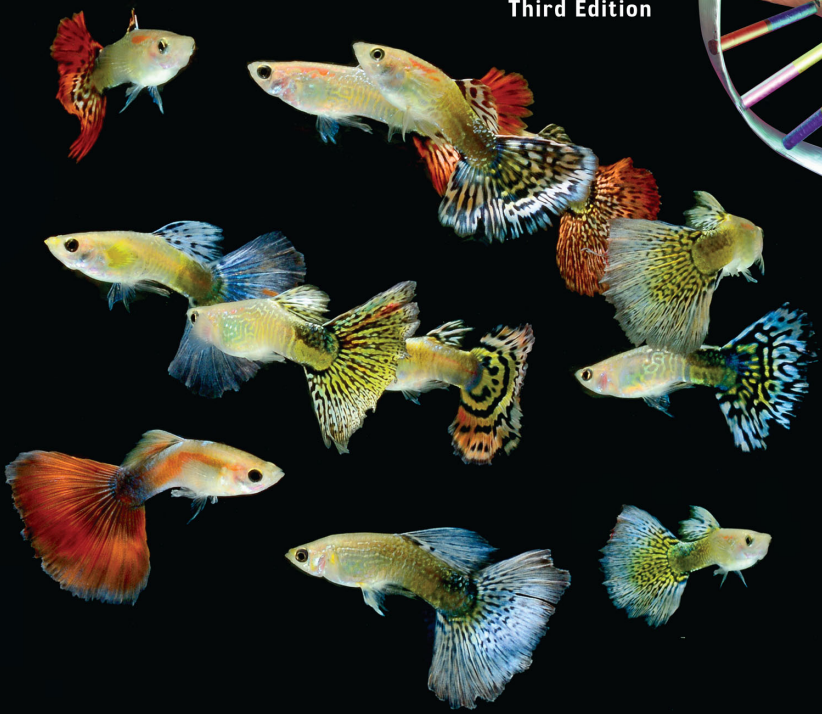
CONCEPTS OF GENETICS

Third Edition



Robert J. Brooker

Mc Graw Hill Education

CONCEPTS OF GENETICS

Third Edition

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CONCEPTS OF GENETICS, THIRD EDITION

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ABOUT THE AUTHOR

Robert J. Brooker is a professor in the Department of Genetics, Cell Biology, and Development and the Department of Biology Teaching and Learning at the University of Minnesota–Minneapolis. He received his B.A. in biology from Wittenberg University in 1978 and his Ph.D. in genetics from Yale University in 1983. At Harvard, he conducted postdoctoral studies on the lactose permease, which is the product of the *lacY* gene of the *lac* operon. He continued his work on transporters at the University of Minnesota. Dr. Brooker's laboratory has also investigated the structure, function, and regulation of manganese and iron transporters found in bacteria and *C. elegans*. At the University of Minnesota, he teaches undergraduate courses in biology, genetics, and cell biology.

Courtesy of Robert Brooker

DEDICATION

To my wife, Deborah, and our children,
Daniel, Nathan, and Sarah

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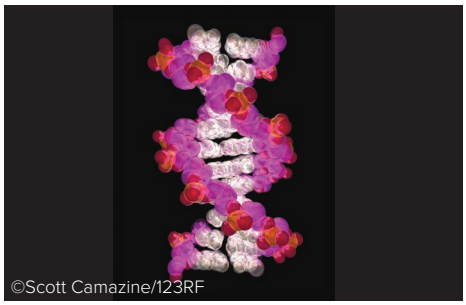
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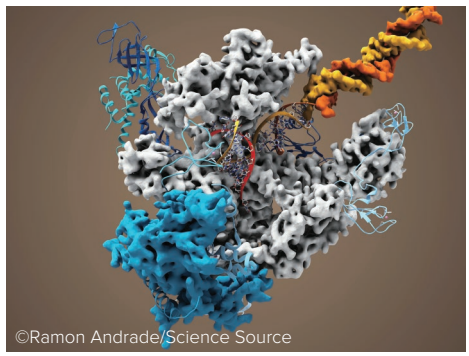
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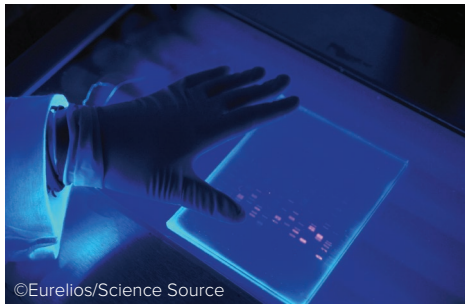
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PREFACE

Based on discussions with instructors from many institutions, I have learned that most instructors want a broad textbook that clearly explains concepts in a way that is interesting, accurate, concise, and up-to-date. *Concepts of Genetics* has been written to achieve these goals. It is intended for students who want to gain a conceptual grasp of the various fields of genetics. The content reflects current trends in genetics, and the pedagogy is based on educational research. In particular, a large amount of formative assessment is woven into the content. As an author, researcher, and teacher, I want a textbook that gets students actively involved in learning genetics. To achieve this goal, I have worked with a talented team of editors, illustrators, and media specialists who have helped me to make the third edition of *Concepts of Genetics* a fun learning tool.

FLIPPING THE CLASSROOM

A trend in science education is the phenomenon that is sometimes called "flipping the classroom." This phrase refers to the idea that some of the activities that used to be done in class are now done out of class, and vice versa. For example, instead of spending the entire class time lecturing about the textbook and other materials, some of the class time is spent engaging students in various activities, such as problem solving, working through case studies, and designing experiments. This approach is called active learning. For many instructors, the classroom has become more learner centered rather than teacher centered. A learner-centered classroom provides a rich environment in which students can interact with each other and with their instructors. Instructors and fellow students often provide formative assessment—immediate feedback that helps students understand if their learning is on the right track.

What are some advantages of active learning? Educational studies reveal that active learning usually promotes greater learning gains. In addition, active learning often focuses on skill development rather than the memorization of facts that are easily forgotten. Students become trained to "think like scientists" and to develop a skill set that enables them to apply scientific reasoning.

A common concern among instructors who are beginning to try out active learning is that they think they will have to teach their students less material. However, this may not be the case. Although students may be provided with online lectures, "flipping the classroom" typically gives students more responsibility for understanding the textbook material on their own. Along these lines, *Concepts of Genetics,* Third Edition, is intended to provide students with a resource that can be effectively used out of the classroom. Several key pedagogical features include the following:

- **Learning Outcomes** Each section of every chapter begins with a set of learning outcomes. These outcomes help students understand what they should be able to do if they have mastered the material in that section.
- **Formative Assessment** When students are expected to learn textbook material on their own, it is imperative that they are given formative assessment on a regular basis so they can gauge whether or not they are mastering the material. Formative assessment is a major feature of this textbook and is bolstered by McGraw-Hill Connect®—a state-of-the art digital assignment and assessment platform. In *Concepts of Genetics,* Third Edition, formative assessment is provided in multiple ways.
    1. Each section of every chapter ends with multiple-choice questions. Formative assessment at the end of each section allows students to evaluate their mastery of the material before moving on to the next section.
    2. Most figures have concept check questions so students can determine if they understand the key points in the figure.
    3. Extensive end-of-chapter questions continue to provide students with feedback regarding their mastery of the material.
    4. A feature called ***Genetic TIPS*** provides a consistent approach to help students solve problems in genetics. This approach has three components: First, the student is made aware of the ***T***opic at hand. Second, the question is evaluated with regard to the ***I***nformation that is available to the student. Finally, the student is guided through a ***P***roblem-***S***olving ***S***trategy to tackle the question.
    5. Additional questions, including questions that pertain to every feature investigation, are available to the student in Connect: http://successinhighered.com/genetics-molecular-biology.
    6. The textbook material is supported by digital learning tools found in Connect. Questions and activities are assignable in Connect. Assignments due before class time or following an in-class activity help students prepare or review.
    7. McGraw-Hill SmartBook is an adaptive learning tool available in Connect that has been shown to strengthen recall and increase retention so that students can move beyond memorizing and truly learn the material.

- **Chapter Organization** In genetics, it is sometimes easy to "lose the forest for the trees." Genetics can be a dense subject. To circumvent this difficulty, the content in *Concepts of Genetics* has been organized to foster a better appreciation for the big picture of genetic principles. The chapters are divided into several sections, and each section ends with a summary that touches on the main points. As mentioned, multiple-choice questions at the end of each section are also intended to help students grasp the broader concepts in genetics. Finally, the end of each chapter contains a summary, which allows students to connect the concepts that were learned in each section.
- **Interactive Exercises** Working with education specialists, the author has crafted interactive exercises in which the students can make their own choices in problem-solving activities and predict what the outcomes will be. Many of these exercises are focused on inheritance patterns and human genetic diseases. (For example, see Chapters 5 and 22.) In addition, there are many interactive exercises for the molecular chapters. These types of exercises engage students in the learning process. The interactive exercises are found online, and the corresponding material in the chapter is indicated with an Interactive Exercise icon.

- **Animations** Our media specialists have created over 50 animations for a variety of genetic processes. These animations were made specifically for this textbook and use the art from the textbook. The animations literally make many of the figures in the textbook "come to life." The animations are found online and the corresponding material in the chapter is indicated with an Online Animation icon.

An effective textbook needs to accomplish three goals: First, it needs to provide comprehensive, accurate, and up-to-date content in its field. Second, it needs to expose students to the techniques and skills they will need to become successful in that field. And finally, it should inspire students so they want to study the material. The hard work that has gone into the third edition of *Concepts of Genetics* has been aimed at achieving all three of these goals. Furthermore, the pedagogy of *Concepts of Genetics* has been designed to foster student learning. Instead of being a collection of "facts and figures," *Concepts of Genetics,* Third Edition, by Robert Brooker, is intended to be an engaging and motivating textbook in which formative assessment allows students to move ahead and learn the material in a productive way. We welcome your feedback so we can make future editions even better!

HOW WE EVALUATED YOUR NEEDS

ORGANIZATION

In surveying many genetics instructors, it became apparent that most people fall into two camps: **Mendel first** versus **Molecular first.** I have taught genetics both ways. As a teaching tool, this textbook has been written with these different teaching strategies in mind. The organization and content lend themselves to various teaching formats.

Chapters 2 through 10 are largely inheritance chapters, whereas Chapters 23 and 24 examine population and quantitative genetics. The bulk of the molecular genetics is found in Chapters 11 through 22, although I have tried to weave a fair amount of molecular genetics into Chapters 2 through 10 as well. The information in Chapters 11 through 22 does not assume that a student has already covered Chapters 2 through 10. Actually, each chapter is written with the perspective that instructors may want to vary the order of their chapters to fit their students' needs.

For those who like to discuss inheritance patterns first, a common strategy would be to cover Chapters 1 through 10 first, and then possibly 23 and 24. (However, many instructors like to cover quantitative and population genetics at the end. Either way works fine.) The more molecular and technical aspects of genetics would then be covered in Chapters 11 through 22. Alternatively, if you like the "Molecular first" approach, you would probably cover Chapter 1, then skip to Chapters 11 through 22, then return to Chapters 2 through 10, and then cover Chapters 23 and 24 at the end of the course. This textbook was written in such a way that either strategy works well.

ACCURACY

Both the publisher and I acknowledge that inaccuracies can be a source of frustration for both the instructor and students. Therefore, throughout the writing and production of this textbook we have worked very hard to catch and correct errors during each phase of development and production.

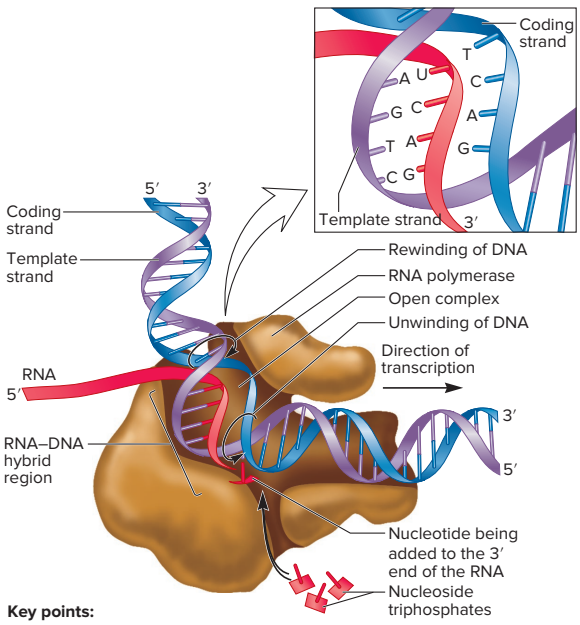
Each chapter has been reviewed by faculty members who teach the course or conduct research in genetics or both. In addition, a developmental editor has gone through the material to check for accuracy in art and consistency between the text and art. When the problem sets were first developed, we had a team of students work through all of the problems and one developmental editor also checked them. The author personally checked every question and answer when the chapters were completed for this edition.

ILLUSTRATIONS

In surveying students whom I teach, I often hear it said that most of their learning comes from studying the figures. Likewise, instructors frequently use the illustrations from a textbook as a central teaching tool. For these reasons, a great amount of effort has gone into the illustrations. The illustrations are created with four goals in mind:

1. **Completeness** For most figures, it should be possible to understand an experiment or genetic concept by looking at the illustration alone. Students have complained that it is difficult to understand the content of an illustration if they have to keep switching back and forth between the figure and text. In cases where an illustration shows the steps in a scientific process, the steps are described in brief statements that allow the students to understand the whole process (e.g., see Figure 17.10). Likewise, such illustrations should make it easier for instructors to explain these processes in the classroom.

2. **Clarity** The figures have been extensively reviewed by students and instructors. This has helped us to avoid drawing things that may be confusing or unclear. Aside from being unmistakably drawn, all new elements within each figure are clearly labeled.
3. **Consistency** Before we began to draw the figures, we generated a style sheet that contained recurring elements that are found in many places in the textbook. Examples include the DNA double helix, DNA polymerase, and fruit flies. We agreed on the best way(s) to draw these elements and also what colors they should be. Therefore, as students and instructors progress through this textbook, they become accustomed to the way things should look.
4. **Realism** An important emphasis of this textbook is to make each figure as realistic as possible. When drawing macroscopic elements (e.g., fruit flies, pea plants), the illustrations are based on real images, not on cartoonlike simplifications. Our most challenging goal, and one that we feel has been achieved most successfully, is the realism of our molecular drawings. Whenever possible, we have tried to depict molecular elements according to their actual structures, if such structures are known. For example, the ways we have drawn RNA polymerase, DNA polymerase, DNA helicase, and ribosomes are based on their crystal structures. When a student sees a figure in this textbook that illustrates an event in transcription, RNA polymerase is depicted in a way that is as realistic as possible (e.g., Figure 14.8 below).



Key points:

- RNA polymerase slides along the DNA, creating an open complex as it moves.
- The DNA strand known as the template strand is used to make a complementary copy of RNA, resulting in an RNA–DNA hybrid.
- RNA polymerase moves along the template strand in a 3′ to 5′ direction, and RNA is synthesized in a 5′ to 3′ direction using nucleoside triphosphates as precursors. Pyrophosphate is released (not shown).
- The complementarity rule is the same as the AT/GC rule except that U is substituted for T in the RNA.

FEATURE EXPERIMENTS

Many chapters have one or two experiments that are presented according to the scientific method. These experiments are integrated within the chapters and flow with the rest of the text. As you are reading the experiments, you will simultaneously explore the scientific method and the genetic principles that have been discovered using this approach. For students, I hope this textbook helps you to see the fundamental connection between scientific analysis and principles. For both students and instructors, I expect that this strategy makes genetics much more fun to explore.

WRITING STYLE

Motivation in learning often stems from enjoyment. If you enjoy what you're reading, you are more likely to spend longer amounts of time with it and focus your attention more crisply. The writing style of this book is meant to be interesting, down to earth, and easy to follow. Each section of every chapter begins with an overview of the contents of that section, usually with a table or figure that summarizes the broad points. The section then examines how those broad points were discovered experimentally, as well as explaining many of the finer scientific details. Important terms appear in the text in a boldface font. These terms are also found at the end of the chapter and in the glossary.

There are various ways to make a genetics book interesting and inspiring. The subject matter itself is pretty amazing, so it's not difficult to build on that. In addition to describing the concepts and experiments in ways that motivate students, it is important to draw on examples that bring the concepts to life. In a genetics book, many of these examples come from the medical realm. This textbook contains lots of examples of human diseases that convey some of the underlying principles of genetics. Students often say they remember certain genetic concepts because they remember how defects in certain genes can cause disease. For example, defects in DNA repair genes cause a higher predisposition to develop cancer. In addition, I have tried to be evenhanded in providing examples from the microbial and plant world. Finally, students are often interested in applications of genetics that affect their everyday lives. Because we frequently hear about genetics in the news, it's inspiring for students to learn the underlying basis for such technologies. Chapters 20 and 21 are devoted to genetic technologies, and applications of these and other technologies are found throughout this textbook. By the end of their genetics course, students should come away with a greater appreciation for the influence of genetics in their lives.

SIGNIFICANT CONTENT CHANGES TO THE THIRD EDITION

- A new feature called ***Genetic TIPS*** was added to the second edition. Many of these problem-solving activities have been refined based on student and instructor feedback.

Examples of Specific Content Changes to Individual Chapters

In addition to the usual updates to material based on new research information, other additions and changes to the third edition have been made, as described next.

- Chapter 5. Extensions of Mendelian Inheritance: A new section has been added that explores how development plays a role in producing certain traits. In particular, having dark fur on the back and white fur on the underside, a trait that is observed in certain breeds of dogs and other animals, is discussed in the context of the migration of melanocyte precursor cells during embryonic development (see Figure 5.13).
- Chapter 7. Genetic Linkage and Mapping in Eukaryotes: Based on student feedback, the discussion of linkage has been improved by increased emphasis on the description of how crossing over is related to the production of recombinant offspring and by the revision of certain figures (e.g., Figure 7.8).
- Chapter 10. Genetics of Viruses: New information about Zika virus has been added.
- Chapter 12. Molecular Structure of Chromosomes and Transposition: The discussion of the mechanism of bacterial chromosome compaction has been updated with a new figure (see Figure 12.3). The topic of transposable elements has been moved to this chapter because of their impact on chromosome structure and genome size.
- Chapter 13. DNA Replication and Recombination: Based on instructor feedback, the topic of homologous recombination has been added to this chapter so that it appears before many of the molecular topics in subsequent chapters.
- Chapter 14. Gene Transcription and RNA Modification: The topic of alternative splicing has been moved to this chapter so that it immediately follows the discussion of splicing.
- Chapter 17. Gene Regulation in Eukaryotes: The discussion of epigenetics has been expanded from one to three sections, which begins with an overview and then focuses on how epigenetic changes are programmed during development and how environmental factors cause epigenetic changes.
- *NEW!* Chapter 18. Non-Coding RNAs: In the past decade or so, technical advances have allowed researchers to identify and study the functions of RNA molecules that do not encode proteins. This new chapter has been added to this edition to focus on this critical topic in molecular genetics.
- Chapter 20. Molecular Technologies: Discussion of CRISPR-Cas technology for gene editing has been added to this chapter (see Figure 20.18), and the topics of reproductive cloning and stem cells have been moved to this chapter from a later chapter.
- Chapter 21. Genomics: The coverage of the topics of genomics and functional genomics has been streamlined and combined in this chapter. The relatively new method of RNA sequencing (RNA-Seq) has been added (see Figure 21.14).
- Chapter 22. Medical Genetics and Cancer: This chapter now covers the use of genome-wide association studies (GWAS) to identify genes associated with human diseases (see Figure 22.8). The section on the genetic basis of cancer has been subdivided into four sections: 22.4 Overview of Cancer, 22.5 Oncogenes, 22.6 Tumor-Suppressor Genes, and 22.7 Role of Epigenetics and Cancer.
- Chapter 24. Quantitative Genetics: The discussion of identifying genes involved in quantitative traits has been set apart in its own section.

SUGGESTIONS WELCOME!

It seems very appropriate to use the word *evolution* to describe the continued development of this textbook. I welcome any and all comments. The refinement of any science textbook requires input from instructors and their students. These include comments regarding writing, illustrations, supplements, factual content, and topics that may need greater or less emphasis. You are invited to contact me at:

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>
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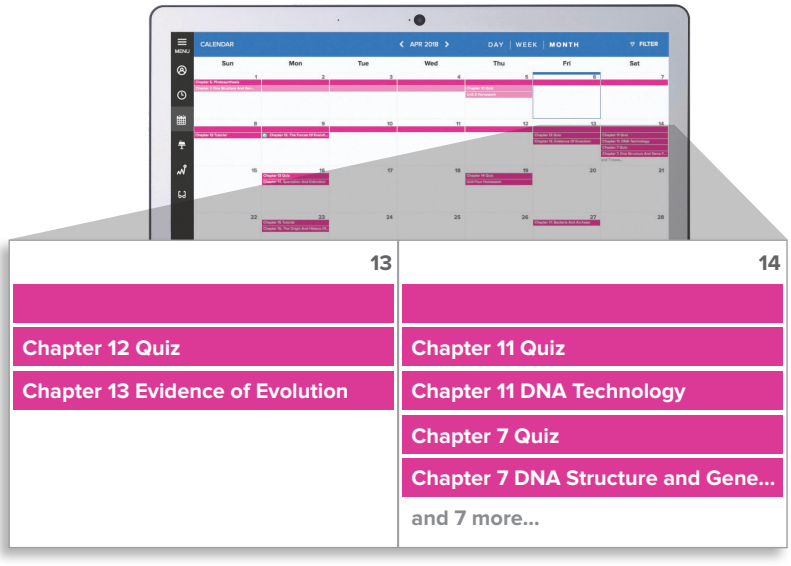

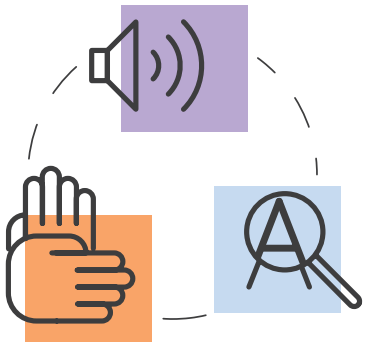

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ACKNOWLEDGMENTS

The production of a textbook is truly a collaborative effort, and I am greatly indebted to a variety of people. This textbook has gone through multiple rounds of rigorous revision that involved the input of faculty, students, editors, and educational and media specialists. Their collective contributions are reflected in the final outcome.

Let me begin by acknowledging the many people at McGraw-Hill Education whose efforts are amazing. My highest praise goes to Andrew Urban (Portfolio Manager), who managed many aspects of this project. I also would like to thank Elizabeth Sievers (Senior Product Developer) for her patience in overseeing this project and her contributions to the digital components. Liz is the glue that holds textbook development and textbook production together. Other people at McGraw-Hill have played key roles in producing an actual book and the supplements that go along with it. In particular, Jessica Portz (Content Project Manager) has done a superb job of managing the components that need to be assembled to produce a book. I would also like to thank Lori Hancock (Content Licensing Specialist), who acted as an interface between me and the photo company. In addition, my gratitude goes to David Hash (Designer), who provided much input into the internal design of the book as well as creating an awesome cover. Finally, I would like to thank Kelly Brown (Marketing Manager), whose major efforts begin once the book is published!

With regard to the content of the book, Deborah Brooker (Freelance Developmental Editor) has worked closely with me in developing the art for this textbook for all of the editions. She has scrutinized each figure for clarity and logic. I would also like to thank Jane Hoover (Freelance Copy Editor) for her superb copy editing. Her crisp understanding of the material allows her to edit it in a meaningful way, which has significantly improved the text's clarity.

I would also like to extend my thanks to everyone at MPS Limited, including the many artists who have played important roles in developing the art for this textbook. Also, the folks at MPS Limited worked with great care in the paging of the book, making sure that the figures and relevant text are as close to each other as possible. Likewise, the people at Aptara have done a great job of locating many of the photographs that have been used in this textbook.

Finally, I want to thank the many scientists who reviewed the chapters of this textbook and previous editions. Their broad insights and constructive suggestions were an important factor that shaped its final content and organization. I am truly grateful for their time and effort.

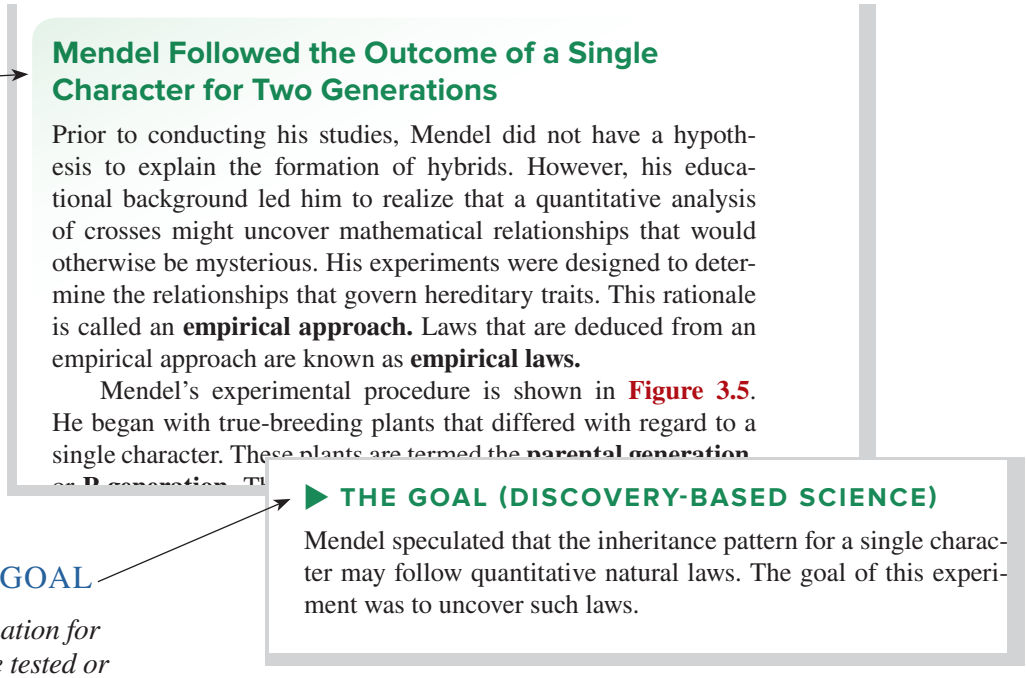
A Visual Guide to
CONCEPTS OF GENETICS

LEARNING THROUGH EXPERIMENTATION

Many chapters contain an experiment that is presented according to the scientific method. These experiments are integrated within the chapters and flow with the rest of the chapter material. As you read the experiments, which can be hypothesis-testing or discovery-based science, you will simultaneously explore the scientific method and the genetic principles learned from this approach.

BACKGROUND OBSERVATIONS

Each experiment begins with a description of the information that led researchers to study a hypothesis-driven or discovery-based problem. Detailed information about the researchers and the experimental challenges they faced help students to understand actual research.



Mendel Followed the Outcome of a Single Character for Two Generations

Prior to conducting his studies, Mendel did not have a hypothesis to explain the formation of hybrids. However, his educational background led him to realize that a quantitative analysis of crosses might uncover mathematical relationships that would otherwise be mysterious. His experiments were designed to determine the relationships that govern hereditary traits. This rationale is called an **empirical approach.** Laws that are deduced from an empirical approach are known as **empirical laws.**

Mendel's experimental procedure is shown in **Figure 3.5**. He began with true-breeding plants that differed with regard to a single character. These plants are termed the **parental generation**

▶ THE GOAL (DISCOVERY-BASED SCIENCE)

Mendel speculated that the inheritance pattern for a single character may follow quantitative natural laws. The goal of this experiment was to uncover such laws.

THE HYPOTHESIS OR THE GOAL

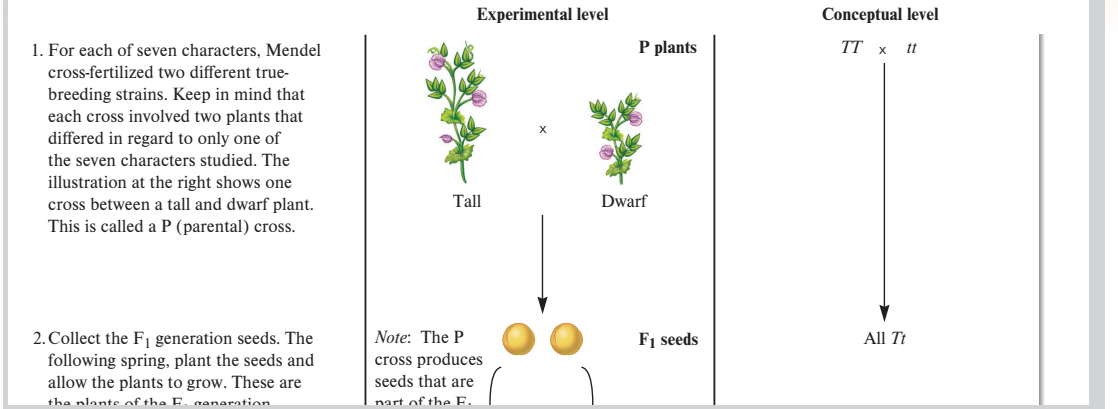
The student is given a possible explanation for the observed phenomenon that will be tested or the question researchers were hoping to answer. This section reinforces the scientific method and allows students to experience the process for themselves.

▶ THE GOAL (DISCOVERY-BASED SCIENCE)

Mendel speculated that the inheritance pattern for a single character may follow quantitative natural laws. The goal of this experiment was to uncover such laws.

▶ ACHIEVING THE GOAL — FIGURE 3.5 Mendel's analysis of single-factor crosses.

Starting material: Mendel began his experiments with true-breeding pea plants that varied with regard to only one of seven different characters (see Figure 3.4).



TESTING THE HYPOTHESIS OR ACHIEVING THE GOAL

This section illustrates the experimental process, including the actual steps followed by scientists to test their hypothesis or study a question. Science comes alive for students with this detailed look at experimentation.

THE DATA

Actual data from the original research paper help students understand how real-life research results are reported. Each experiment's results are discussed in the context of the larger genetic principle to help students understand the implications and importance of the research.

▶ THE DATA

P cross	*F_1 generation*	*F_2 generation*	*Ratio of traits in F_2 generation*
Tall × dwarf height	All tall	787 tall, 277 dwarf	2.84:1
Purple × white flowers	All purple	705 purple, 224 white	3.15:1
Axial × terminal flowers	All axial	651 axial, 207 terminal	3.14:1
Yellow × green seeds	All yellow	6022 yellow, 2001 green	3.01:1
Round × wrinkled seeds	All round	5474 round, 1850 wrinkled	2.96:1
Green × yellow pods	All green	428 green,	

INTERPRETING THE DATA

This discussion, which examines whether the experimental data supported or disproved the hypothesis or provided new information to propose a hypothesis, gives students an appreciation for scientific interpretation.

▶ INTERPRETING THE DATA

The data shown in the table above are the results of producing an F_1 generation via cross-fertilization and an F_2 generation via self-fertilization of the F_1 monohybrids. A quantitative analysis of these data allowed Mendel to propose three important ideas:

1. Mendel's data argued strongly against a blending mechanism of inheritance. In all seven cases, the F_1 generation displayed traits that were distinctly like one of the two parents rather than traits intermediate in character. His first proposal was that one variant of a character is **dominant** to another variant. For example, the variant of green pods is dominant to that of yellow pods. The term **recessive** is used to describe a variant that is masked by the presence of a dominant trait but reappears in subsequent generations. Yellow pods and dwarf height are examples of recessive variants. They can also be referred to as recessive traits.
2. When a true-breeding plant with a dominant trait was crossed to a true-breeding plant with a recessive trait, the dominant trait was always observed in the F_1 generation. In the F_2 generation, some offspring displayed the dominant trait, but a smaller proportion showed the recessive trait. How did Mendel explain this observation? Because the recessive trait appeared in the F_2 generation, he made a second proposal—the genetic determinants of traits are passed along as "unit factors" from generation to generation. His data were consistent with a **particulate theory of inheritance,** in which the genes that govern traits are inherited as discrete units that remain unchanged as they are passed from parent to offspring. Mendel called them unit factors, but we now call them genes (from the Greek, *genesis*, meaning "birth," or *genos*, meaning "origin").

Learning–Assessment–Problem Solving

These study tools and problems are crafted to aid students in reviewing key information in the text, assess their understanding, and develop problem-solving skills.

18.1 OVERVIEW OF NON-CODING RNAs

Learning Outcomes:

1. Describe the ability of ncRNAs to bind to other molecules and macromolecules.
2. Outline the general functions of ncRNAs.
3. Define *ribozyme*.
4. List several examples of ncRNAs, and describe their functions.

LEARNING OUTCOMES

Each section begins with one or more Learning Outcomes. These allow a student to appreciate the skills and knowledge they will gain if they master the material.

REVIEWING THE KEY CONCEPTS

These bulleted lists at the end of each section help students identify important concepts. Students should understand these concepts before moving on to the next section.

COMPREHENSION QUESTIONS

Multiple choice questions found at the end of each section allow students an opportunity to test their knowledge of key information and concepts. This helps students better identify what they know and don't know before tackling more concepts. Answers are provided at the end of the chapter.

4.2 REVIEWING THE KEY CONCEPTS

- Dosage compensation often occurs in species in which males and females differ in their sex chromosomes (see Table 4.1).
- In mammals, the process of X-chromosome inactivation (XCI) in females compensates for the single X chromosome found in males. The inactivated X chromosome is called a Barr body. The process can lead to a variegated phenotype, such as a calico cat (see Figure 4.5).
- After it occurs during embryonic development, X-chromosome inactivation is maintained when somatic cells divide (see Figure 4.6).
- X-chromosome inactivation is controlled by the X-inactivation center (Xic) that contains the *Xist* gene. The three phases of XCI are initiation, spreading, and maintenance phases (see Figure 4.7).

4.2 COMPREHENSION QUESTIONS

1. In fruit flies, dosage compensation is achieved by
   a. X-chromosome inactivation.
   b. doubling the expression of genes on the single X chromosome in the male.
   c. decreasing the expression of genes on each X chromosome in the female to one-half.
   d. all of the above.
2. According to the Lyon hypothesis,
   a. one of the X chromosomes is converted to a Barr body in somatic cells of female mammals.
   b. one of the X chromosomes is converted to a Barr body in all cells of female mammals.
   c. both of the X chromosomes are converted to Barr bodies in somatic cells of female mammals.
   d. both of the X chromosomes are converted to Barr bodies in all cells of female mammals.
3. Which of the following is *not* a phase of XCI?
   a. Initiation
   b. Spreading
   c. Maintenance
   d. Erasure

GENES → TRAITS

Because genetics is such a broad discipline ranging from the molecular level to populations, many students have trouble connecting the concepts they learn in molecular genetics with the traits that occur at the level of an organism. To make this connection more meaningful, certain figures have a "Genes→Traits" feature that reminds students that molecular and cellular phenomena ultimately lead to traits observed in organisms.

CONCEPT CHECK QUESTIONS

Students can test their knowledge and understanding with Concept Check questions that are associated with the figure legends. These questions often go beyond simple recall of information and ask students to apply or interpret information presented in the illustrations.

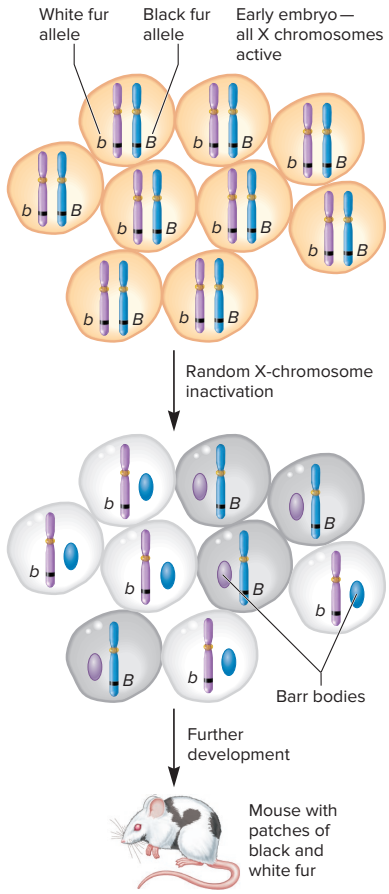


FIGURE 4.6 **The mechanism of X-chromosome inactivation.**

Genes → Traits The top of this figure represents a mass of several cells that compose the early embryo. Initially, both X chromosomes are active. At an early stage of embryonic development, random inactivation of one X chromosome occurs in each cell. This inactivation pattern is maintained as the embryo matures into an adult.

Concept Check: *At which stage of development does X-chromosome inactivation initially occur?*

Genetic TIPS

The Question: If a diploid cell contains four chromosomes (i.e., two per set), how many possible random arrangements of homologs could occur during metaphase of meiosis I?

Topic: What topic in genetics does this question address?

The topic is meiosis. More specifically, the question is about metaphase of meiosis I.

Information: What information do you know based on the question and your understanding of the topic?

From the question, you know a cell that started with two pairs of homologous chromosomes has entered meiosis and is now in metaphase of meiosis I. From your understanding of the topic, you may remember that tetrads align along the metaphase plate (see Figure 2.11). The orientations of the homologs within the tetrads are random.

Problem-Solving Strategies: Make a drawing. Make a calculation.

One strategy to solve this problem is to make a drawing in which the homologs are in different colors, such as red and blue. Note: The spindle poles are labeled A and B in the drawing below. The alignment occurs relative to the spindle poles.

GENETIC TIPS

*Problem solving is a skill that genetics students need to master. **Genetic TIPS** provides a consistent approach to help students solve problems in genetics. This approach has three components: First, the student is made aware of the **T**opic at hand. Second, the question is evaluated with regard to the **I**nformation that is available to the student. Finally, the student is guided through a **P**roblem-Solving **S**trategy to tackle the question. **More Genetic TIPS** are presented at the end of the chapter, allowing for additional practice in strengthening problem-solving skills.*

End-of-Chapter Support Materials

These study tools and problems are crafted to aid students in reviewing key information in the text and developing a wide range of problem-solving skills. They also develop a student's cognitive, writing, analytical, computational, and collaborative abilities.

KEY TERMS

Providing the key terms from the chapter enhances student development of vital vocabulary necessary for the understanding and application of chapter content. Important terms are boldfaced throughout the chapter and page referenced at the end of each chapter for reflective study.

CHAPTER SUMMARY

These bulleted summaries, which are organized by section, emphasize the main concepts of the chapter to provide students with a thorough review of the main topics covered.

KEY TERMS

Page 114. nuclear genes, extranuclear inheritance (cytoplasmic inheritance), nucleoid, chloroplast DNA (cpDNA)
Page 115. reciprocal cross, maternal inheritance, heteroplasmy
Page 116. heterogamous
Page 117. mitochondrial DNA (mtDNA)
Page 118. paternal leakage
Page 119. endosymbiosis, endosymbiosis theory
Page 120. epigenetic inheritance, genomic imprinting (imprinting), monoallelic expression
Page 122. DNA methylation, imprinting control region (ICR)
Page 124. maternal effect

CHAPTER SUMMARY

- Non-Mendelian inheritance refers to inheritance patterns that do not obey Mendel's laws of inheritance.

6.1 Extranuclear Inheritance: Chloroplasts

- Extranuclear inheritance is the inheritance of genes that are found in chloroplasts or mitochondria.
- Chloroplasts have circular chromosomes in a nucleoid. These circular chromosomes carry many genes but far fewer compared with the number on chromosomes in the cell nucleus (see Figure 6.1, Table 6.1).
- Maternal inheritance occurs when organelles, such as chloroplasts, are transmitted via the egg (see Figure 6.2).
- Heteroplasmy of chloroplasts can result in a variegated pheno-

6.3 Theory of Endosymbiosis

- Chloroplasts and mitochondria were derived from ancient endosymbiotic relationships (see Figure 6.6).

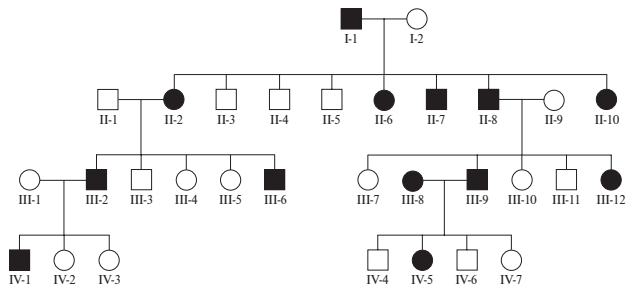
6.4 Epigenetics: Imprinting

- Epigenetic inheritance is an inheritance pattern in which a gene or chromosome is modified so as to alter gene expression, but the modification is not permanent over the course of many generations. An example is imprinting, in which an offspring expresses a gene that is inherited from one parent but not both (see Figures 6.7, 6.8).
- DNA methylation at an imprinting control region is the marking

PROBLEM SETS & INSIGHTS

More Genetic TIPS

1. The pedigree presented here shows the inheritance of a human disease known as familial hypercholesterolemia.



This disorder is characterized by an elevated level of serum cholesterol in the blood. Though relatively rare, this genetic abnormality

Information: What information do you know based on the question and your understanding of the topic?

MORE GENETIC TIPS

*Like the Genetic TIPS within the chapter, these problems provide more practice in developing problem-solving skills before the students work through more problems unaided. The Genetic TIPS help the student identify the primary question (the **T**opic), evaluate the question based on the student's knowledge of the topic (**I**nformation), and then the student is guided through the solution revealing a **P**roblem-Solving **S**trategy. These provide a reference for when students encounter similar problems later.*

CONCEPTUAL QUESTIONS

These questions test the understanding of basic genetic principles. The student is given many questions with a wide range of difficulty. Some require critical-thinking skills, and some require the student to write coherent answers in an essay form.

Conceptual Questions

C1. The process of binary fission begins with a single mother cell ends with two daughter cells. Would you expect the mother an daughter cells to be genetically identical? Explain why or why not.

C2. What is a homolog? With regard to genes and alleles, how are homologs similar to and different from each other?

C3. What is a sister chromatid? Are sister chromatids genetically s lar or identical? Explain.

C4. With regard to sister chromatids, which phase of mitosis is the organization phase, and which is the separation phase?

C5. A species is diploid with three chromosomes per set. Make a d ing that shows what the chromosomes would look like in the C and G_2 phases of the cell cycle.

C6. How does the attachment of kinetochore microtubules to the k ochore differ in metaphase of meiosis I from metaphase of mit Discuss what you think would happen if a sister chromatid wa attached to a kinetochore microtubule.

C7. For the following events, specify whether they occur during m sis, meiosis I, or meiosis II:

a. Separation of conjoined chromatids within a pair of sister

APPLICATION AND EXPERIMENTAL QUESTIONS

These questions test the ability to analyze data, design experiments, or appreciate the relevance of experimental techniques.

Application and Experimental Questions

E1. Describe three advantages of using pea plants as an experimental organism.

E2. Explain the technical differences between a cross-fertilization experiment and a self-fertilization experiment.

E3. How long did it take Mendel to complete the experiment in Figure 3.5?

E4. For all seven characters described in the data of Figure 3.5, Mendel allowed the F_2 plants to self-fertilize. He found that when F_2 plants with recessive traits were crossed to each other, they always bred true. However, when F_2 plants with dominant traits were crossed, some bred true but others did not. A summary of Mendel's results is shown in the following table.

The Ratio of True-Breeding and Non-True-Breeding Parents of the F_2 Generation

F_2 Parents	True-Breeding	Non-True-Breeding	Ratio
Round	193	372	1:1.93
Yellow	166	353	1:2.13
Gray	36	64	1:1.78
Smooth	29	71	1:2.45
Green	40	60	1:1.5
Axial	33	67	1:2.08
Tall	28	72	1:2.57
TOTAL:	525	1059	1:2.02

When considering the data in this table, keep in mind that they describe the characteristics of the F_2 generation parents that had

QUESTIONS FOR STUDENT DISCUSSION/ COLLABORATION

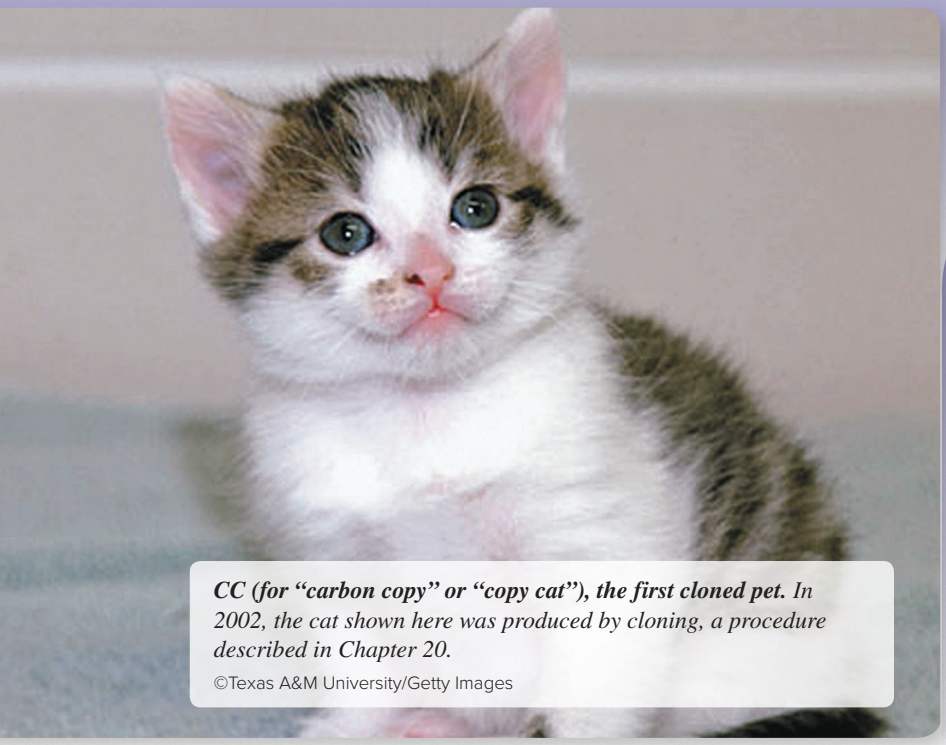
These questions encourage students to consider broad concepts and practical problems. Some questions require a substantial amount of computational activities, which can be worked on as a group.

Questions for Student Discussion/Collaboration

1. Consider this cross in pea plants: *Tt Rr yy Aa* × *Tt rr Yy Aa*, where *T* = tall, *t* = dwarf, *R* = round, *r* = wrinkled, *Y* = yellow, *y* = green, *A* = axial, *a* = terminal. What is the expected phenotypic outcome of this cross? Have one group of students solve this problem by making one big Punnett square, and have another group solve it by making four single-gene Punnett squares and using the multiplication method. Time each other to see who gets done first.

or dwarf with terminal flowers and the fourth offspring will be tall with axial flowers? Discuss what operation(s) (e.g., product rule or binomial expansion equation) you used and in what order they were used.

3. Consider this four-factor cross: *Tt Rr yy Aa* × *Tt RR Yy aa*, where *T* = tall, *t* = dwarf, *R* = round, *r* = wrinkled, *Y* = yellow, *y* = green, *A* = axial, *a* = terminal. What is the probability that the first three plants



CC (for "carbon copy" or "copy cat"), the first cloned pet. *In 2002, the cat shown here was produced by cloning, a procedure described in Chapter 20.*

1

CHAPTER OUTLINE

OVERVIEW OF GENETICS

Hardly a week goes by without a major news story involving a genetic breakthrough. The increasing pace of genetic discoveries has become staggering. The Human Genome Project is a case in point. This project began in the United States in 1990, when the National Institutes of Health and the Department of Energy joined forces with international partners to decipher the massive amount of information contained in our **genome**—the **deoxyribonucleic acid (DNA)** found within all of our chromosomes (**Figure 1.1**). Remarkably, in only a decade, the researchers working on this project determined the DNA sequence of 90% of the human genome. The completed sequence, published in 2003, has an accuracy greater than 99.99%; fewer than one mistake was made in every 10,000 base pairs (bp)!

In 2008, a more massive undertaking, called the 1000 Genomes Project, was launched, with the goal of establishing a detailed understanding of human genetic variation. In this international project, researchers set out to determine the DNA sequence of at least 1000 anonymous participants from around the globe. In 2015, the sequencing of over 2500 genomes was described in the journal *Nature*.

Studying the human genome allows us to explore fundamental details about ourselves at the molecular level. The results of human genome projects have shed considerable light on basic questions, such as how many genes we have, how genes direct the activities of living cells, how species evolve, how single cells develop into complex tissues, and how defective genes cause disease. Furthermore, such understanding may lend itself to improvements in modern medicine by providing better diagnoses of diseases and allowing the development of new treatments for them.

A controversial example of a genetic technology is mammalian cloning. In 1997, Ian Wilmut and his colleagues produced clones of sheep, using mammary cells from an adult animal (**Figure 1.2**). More recently, such cloning has been achieved in several mammalian species, including cows, mice, goats, pigs, and cats. In 2002, the first pet was cloned, a cat named CC (for "carbon copy" or "copy cat"; see the photo at the beginning of the chapter). The cloning of mammals provides the potential for many practical applications. Cloning of livestock would enable farmers to use cells from their best individuals to create genetically homogeneous herds. This could be advantageous in terms of agricultural yield, although such a genetically homogeneous herd may be more susceptible to certain diseases. However, people have become greatly concerned with the possibility of human cloning. As discussed in Chapter 20, this prospect has raised serious ethical questions. Within the past few years, legislative bills have been introduced that involve bans on human cloning.

DNA, the molecule of life

The adult human body is composed of trillions of cells.

Most human cells contain the following:

- 46 human chromosomes, found in 23 pairs
- 2 meters of DNA
- Approximately 22,000 genes coding for proteins that perform most life functions
- Approximately 3 billion DNA base pairs per set of chromosomes, containing the bases A, T, G, and C

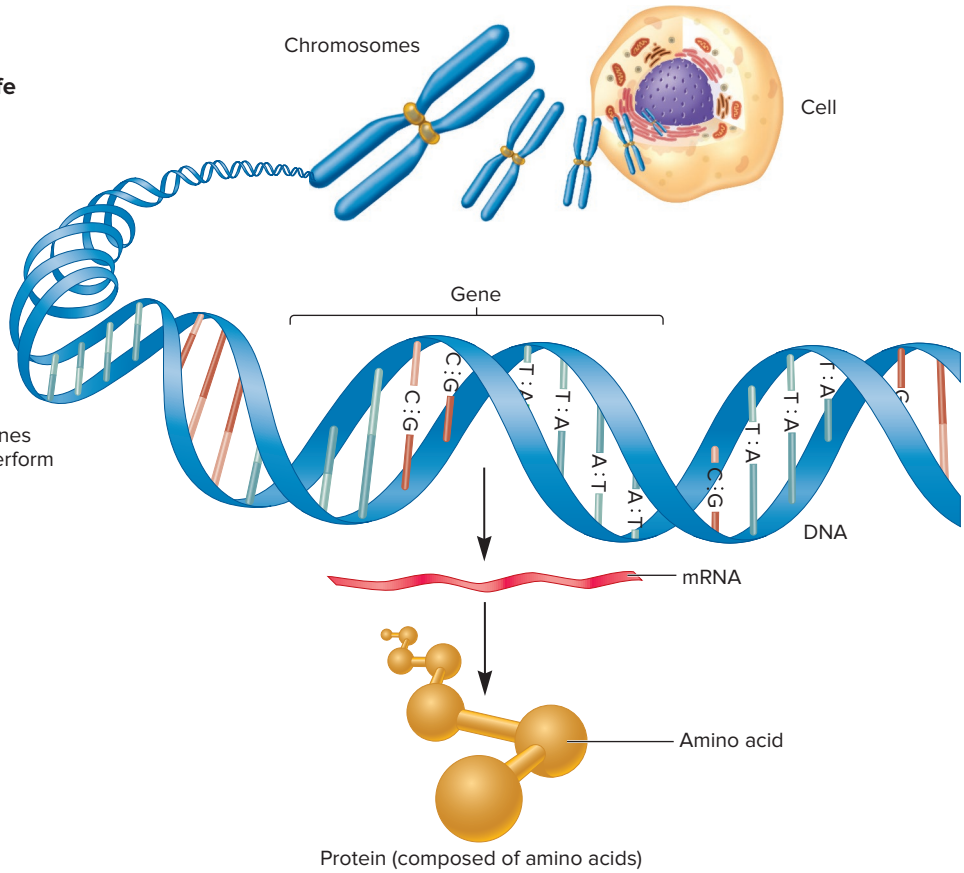


FIGURE 1.1 **The Human Genome Project.** The human genome is a complete set of human chromosomes. People have two sets of chromosomes, one set from each parent. Collectively, each set of chromosomes is composed of a DNA sequence that is approximately 3 billion nucleotide base pairs long. Estimates suggest that each set contains about 22,000 protein-encoding genes. This figure emphasizes the DNA found in the cell nucleus. Humans also have a small amount of DNA in their mitochondria, which has also been sequenced.

Concept Check: *How might a better understanding of our genes be used in the field of medicine?*

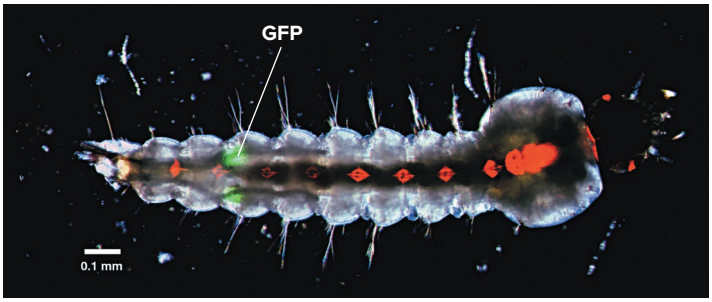
FIGURE 1.2 **The cloning of a mammal.** The lamb on the left is Dolly, the first mammal to be cloned. She was cloned from a cell of a Finn Dorset (a white-faced sheep). The sheep on the right is Dolly's surrogate mother, a Blackface ewe. A description of how Dolly was produced is presented in Chapter 20.

Concept Check: *What ethical issues may be associated with human cloning?*

Finally, genetic technologies provide the means of modifying the traits of animals and plants in ways that would have been unimaginable just a few decades ago. **Figure 1.3a** shows a bizarre example in which scientists introduced a gene from jellyfish into mice. Certain species of jellyfish emit a "green glow" produced by a gene that encodes a bioluminescent protein called green fluorescent protein (GFP). When exposed to blue or ultraviolet (UV) light, the protein emits a striking green-colored light. Scientists were able to clone the *GFP* gene from a sample of jellyfish cells and then introduce this gene into laboratory mice. The green fluorescent protein is made throughout the cells of their bodies. As a result, their skin, eyes, and organs give off an eerie green glow when exposed to UV light.

The expression of green fluorescent protein allows researchers to identify particular proteins in cells or specific body parts. For example, Andrea Crisanti and colleagues have altered mosquitoes to express GFP only in the gonads of males (**Figure 1.3b**). This enables the researchers to distinguish males from females and sort mosquitoes by sex. Why is this useful? The ability to rapidly

(a) GFP expressed in mice



(b) GFP expressed in the gonads of a male mosquito

FIGURE 1.3 The introduction of a jellyfish gene into laboratory mice and mosquitoes. **(a)** A gene that naturally occurs in certain jellyfish encodes a protein called green fluorescent protein (GFP). The *GFP* gene was cloned and introduced into mice. When these mice are exposed to ultraviolet light, GFP emits a bright green color. These mice glow green, just like the jellyfish! **(b)** *GFP* was introduced next to a gene sequence that causes the expression of GFP only in the gonads of male mosquitoes. The resulting green glow allows researchers to identify and sort males from females.

(a) ©Eye of Science/Science Source; (b) Courtesy of Flaminia Catteruccia, Jason Benton and Andrea Crisanti

Concept Check: Why is it useful to sort male mosquitoes from female mosquitoes?

sort mosquitoes by sex makes it possible to produce populations of sterile males and then release the sterile males without the risk of releasing additional females. The release of sterile males may be an effective means of controlling mosquito populations because females breed only once. Mating with a sterile male prevents a female from producing offspring. In 2008, Osamu Shimomura, Martin Chalfie, and Roger Tsien received the Nobel Prize in chemistry for the discovery and the development of GFP, which has become a widely used tool in biology.

Overall, as we move forward in the twenty-first century, the excitement level in the field of genetics is high, perhaps higher than it has ever been. Nevertheless, new genetic knowledge and technologies will also create many ethical and societal challenges. In this chapter, we begin with an overview of genetics and then explore the various fields of genetics and their experimental approaches.

1.1 THE MOLECULAR EXPRESSION OF GENES

Learning Outcomes:

1. Describe the biochemical composition of cells.
2. Outline how DNA stores the information to make proteins.
3. Explain how proteins are largely responsible for cell structure and function.

Genetics is the branch of biology that deals with heredity and variation. It stands as the unifying discipline in biology by allowing us to understand how life can exist at all levels of complexity, ranging from the molecular to the population level. Genetic variation is the root of the natural diversity that we observe among members of the same species and among different species.

Genetics is centered on the study of genes. A gene is classically defined as a unit of heredity, but such a vague definition does not do justice to the exciting characteristics of genes as intricate molecular units that manifest themselves as critical contributors to cell structure and function.

- At the molecular level, a **gene** is a segment of DNA that has the information to produce a functional product. The functional product of most genes is a polypeptide—a linear sequence of amino acids that folds into units that constitute proteins.
- Genes are commonly described according to the way they affect **traits,** which are the characteristics of an organism. In humans, for example, we observe traits such as eye color, hair texture, and height. An ongoing theme of this textbook is the relationship between genes and traits. As an organism grows and develops, its collection of genes provides a blueprint that determines its characteristics.

In this section, we will examine the general features of life with an emphasis on the molecular level. Genetics is the common thread that explains the existence of life and its continuity from generation to generation. For most students, this chapter should serve as a cohesive review of topics they learned in other introductory courses such as general biology. Even so, it is usually helpful to see the "big picture" of genetics before delving into the finer details that are covered in Chapters 2 through 24.

Living Cells Are Composed of Biochemicals

To fully understand the relationship between genes and traits, we need to begin with an examination of the composition of living organisms. Every cell is constructed from intricately organized

chemical substances. Small organic molecules such as glucose and amino acids are produced by the linkage of atoms via chemical bonds. The chemical properties of organic molecules are essential for cell vitality in two key ways.

- First, the breaking of chemical bonds during the degradation of small molecules provides energy to drive cellular processes.
- A second important function of these small organic molecules is their role as the building blocks for the synthesis of larger molecules. Four important categories of larger cellular molecules are **nucleic acids** (i.e., DNA and RNA), **proteins, carbohydrates,** and **lipids.** Three of these—nucleic acids, proteins, and carbohydrates—form **macromolecules** that are composed of many repeating units of smaller building blocks. Proteins, RNA, and carbohydrates can be made from hundreds or even thousands of repeating building blocks. DNA is the largest macromolecule found in living cells. A single DNA molecule can be composed of a linear sequence of hundreds of millions of building blocks called nucleotides!

The formation of cellular structures relies on the interactions of molecules and macromolecules. **Figure 1.4** illustrates this concept.

- Nucleotides are small organic molecules.
- Nucleotides are linked to each other and form the building blocks of DNA, which is a macromolecule.
- DNA is a component of chromosomes, which also contain proteins that contribute to chromosome structure.
- Within a eukaryotic cell, the chromosomes are contained in a compartment called the cell nucleus. The nucleus is bounded by a double membrane composed of lipids and proteins that shields the chromosomes from the rest of the cell. The nucleus is an example of an **organelle**—a membrane-bound compartment with a specialized function. The cell nucleus protects the chromosomes from mechanical damage and provides a single compartment for genetic activities such as gene transcription.
- Finally, cellular molecules, macromolecules, and organelles are organized to make a complete living cell.

Each Cell Contains Many Different Proteins That Determine Cell Structure and Function

To a great extent, the characteristics of a cell depend on the types of proteins that it makes. The entire collection of proteins that a cell makes at a given time is called its **proteome.** As we will learn throughout this textbook, proteins are the "workhorses" of all living cells. The range of functions among different types of proteins is truly remarkable. Some examples include the following:

- Proteins help determine the shape and structure of a given cell. For example, the protein known as tubulin can assemble into large structures known as microtubules, which provide the cell with internal structure and organization.
- Proteins are inserted into cell membranes and aid in the transport of ions and small molecules across the membrane.

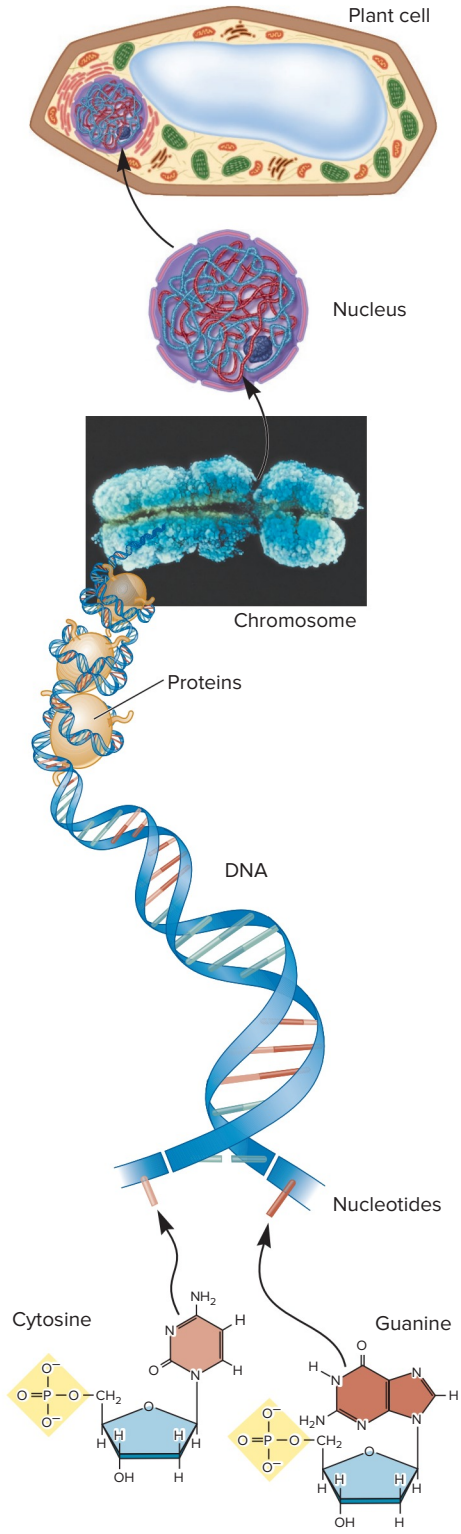


FIGURE 1.4 **Molecular organization of a living cell.** Cellular structures are constructed from smaller building blocks. In this example, DNA is formed from the linkage of nucleotides, producing a very long macromolecule. The DNA associates with proteins to form a chromosome. The chromosomes are located within a membrane-bound organelle called the nucleus, which, along with many different types of organelles, is found within a complete cell.

Concept Check: Is DNA a small molecule, a macromolecule, or an organelle?

- Proteins may also function as biological motors. An interesting case is the protein known as myosin, which is involved in the contractile properties of muscle cells.
- Within multicellular organisms, certain proteins function in cell-to-cell recognition and signaling. For example, hormones such as insulin are secreted by endocrine cells and bind to the insulin receptor protein found within the plasma membrane of target cells.
- **Enzymes,** which accelerate chemical reactions, are a particularly important category of proteins. Some enzymes play a role in the breakdown of molecules or macromolecules into smaller units. These enzymes are important in the utilization of energy.

Molecular biologists have come to realize that the functions of proteins underlie the cellular characteristics of every organism. At the molecular level, proteins can be viewed as the active participants in the enterprise of life.

DNA Stores the Information for Protein Synthesis

As mentioned, the genetic material of living organisms is composed of a substance called deoxyribonucleic acid, abbreviated DNA. The DNA stores the information needed for the synthesis of all proteins. In other words, the main function of the genetic blueprint is to code for the production of proteins in the correct cell, at the proper time, and in suitable amounts. This task is extremely complicated because living cells make thousands of different proteins. Genetic analyses have shown that a typical bacterium can make a few thousand different proteins, and estimates of the numbers of proteins produced by complex eukaryotes range in the tens of thousands.

DNA's ability to store information is based on its structure.

- DNA is composed of a linear sequence of **nucleotides,** each of which contains one of four nitrogen-containing bases: adenine (A), thymine (T), guanine (G), or cytosine (C).
- The linear order of these bases along a DNA molecule contains information similar to the way that groups of letters of the alphabet represent words. For example, the "meaning" of the sequence of bases ATGGGCCTTAGC differs from that of TTTAAGCTTGCC.
- DNA sequences within most genes contain the information to direct the order of amino acids within **polypeptides** according to the **genetic code.** In the code, a three-base sequence, called a **codon,** specifies one particular **amino acid** among the 20 possible choices.
- The sequence of amino acids in a polypeptide causes it to fold into a particular structure; one or more polypeptides form a functional protein.

In this way, the DNA can store the information to specify the proteins made by an organism.

DNA Sequence	Amino Acid Sequence
ATG GGC CTT AGC	Methionine Glycine Leucine Serine
TTT AAG CTT GCC	Phenylalanine Lysine Leucine Alanine

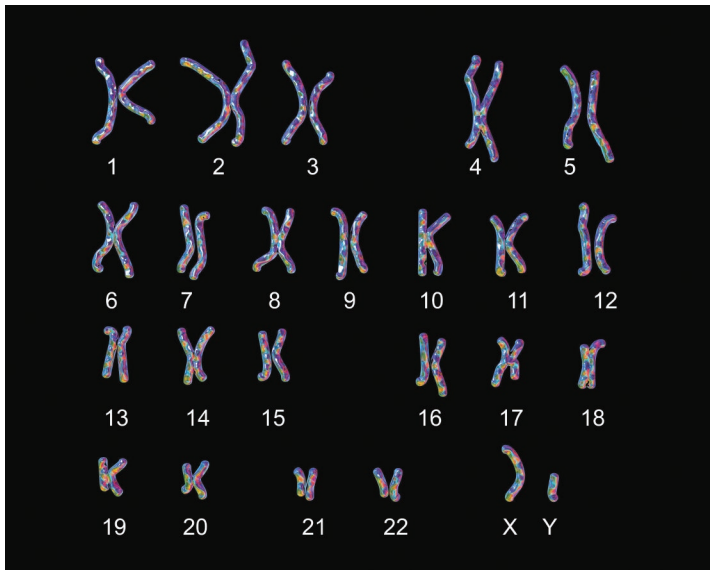


FIGURE 1.5 A micrograph of the 46 chromosomes found in a cell from a human male.

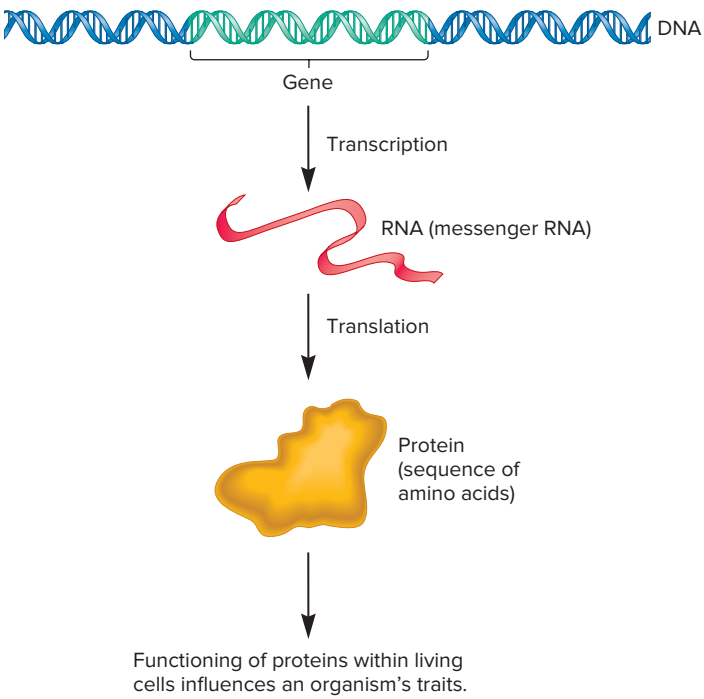
Concept Check: Which types of macromolecules are found in chromosomes?

In living cells, DNA is found within large structures known as **chromosomes.** Figure 1.5 is a micrograph of the 46 chromosomes in a cell from a human male, which are found in pairs. The DNA of an average human chromosome is an extraordinarily long, linear, double-stranded structure that contains well over a hundred million nucleotides. Along the immense length of a chromosome, the genetic information is parceled into functional units known as genes. An average-sized human chromosome is expected to carry about 1000 different genes.

The Information in DNA Is Accessed During the Process of Gene Expression

To synthesize its proteins, a cell must be able to access the information that is stored within its DNA. The process of using a gene sequence to affect the characteristics of cells and organisms is referred to as **gene expression.** At the molecular level, the information is accessed in a stepwise process (Figure 1.6).

1. In the first step, known as **transcription,** the DNA sequence within a gene is copied into a nucleotide sequence of **ribonucleic acid (RNA).** Most genes encode RNAs that contain the information for the synthesis of a particular polypeptide. This type of RNA is called **messenger RNA (mRNA).**
2. During the process of **translation,** the sequence of nucleotides in an mRNA provides the information (using the genetic code) to produce the amino acid sequence of a polypeptide.
3. A polypeptide folds into a three-dimensional structure. As mentioned, a protein is a functional unit. Some proteins are composed of a single polypeptide, and other proteins consist of two or more polypeptides.
4. The functioning of proteins largely determines cell structure and function.



ONLINE ANIMATION

FIGURE 1.6 Gene expression at the molecular level. The expression of a gene is a multistep process. During transcription, one of the DNA strands is used as a template to make an RNA strand. During translation, the RNA strand is used to specify the sequence of amino acids within a polypeptide. One or more polypeptides produce a functional protein, thereby influencing an organism's traits.

Concept Check: Where is the information to make a polypeptide stored?

1.1 REVIEWING THE KEY CONCEPTS

- Living cells are composed of nucleic acids (DNA and RNA), proteins, carbohydrates, and lipids. The proteome largely determines the structure and function of cells (see Figure 1.4).
- DNA, which is found within chromosomes, stores the information to make proteins (see Figure 1.5).
- Most genes encode polypeptides that are units within functional proteins. Gene expression at the molecular level involves transcription to produce mRNA and translation to produce a polypeptide (see Figure 1.6).

1.1 COMPREHENSION QUESTIONS

1. Which of the following is *not* a constituent of a cell's proteome?
   a. An enzyme
   b. A motor protein
   c. A receptor in the plasma membrane
   d. An mRNA
2. A gene is a segment of DNA that has the information to produce a functional product. The functional product of most genes is
   a. DNA.
   b. mRNA.
   c. a polypeptide.
   d. none of the above.
3. The function of the genetic code is to
   a. promote transcription.
   b. specify the amino acids within a polypeptide.
   c. alter the sequence of DNA.
   d. do none of the above.
4. The process of transcription directly results in the synthesis of
   a. DNA.
   b. RNA.
   c. a polypeptide.
   d. all of the above.

1.2 THE RELATIONSHIP BETWEEN GENES AND TRAITS

Learning Outcomes:

1. Outline how the expression of genes leads to an organism's traits.
2. Define *genetic variation*.
3. Discuss the relationship between genes, traits, and the environment.
4. Describe how genes are transmitted in sexually reproducing species.
5. Describe the process of evolution.

A trait is any characteristic that an organism displays. In genetics, we can place traits into different categories.

- **Morphological traits** affect the appearance, form, and structure of an organism. The color of a flower and the height of a pea plant are morphological traits. Geneticists frequently study these types of traits because they are easy to evaluate. For example, an experimenter can simply look at a plant and tell if it has red or white flowers.
- **Physiological traits** affect the ability of an organism to function. For example, the rate at which a bacterium metabolizes a sugar such as lactose is a physiological trait. Like morphological traits, physiological traits are controlled, in part, by the expression of genes.
- **Behavioral traits** affect the ways an organism responds to its environment. An example is the mating calls of bird species. In animals, the nervous system plays a key role in governing such traits.

In this section, we will examine the relationship between the expression of genes and an organism's traits.

The Molecular Expression of Genes Within Cells Leads to an Organism's Traits

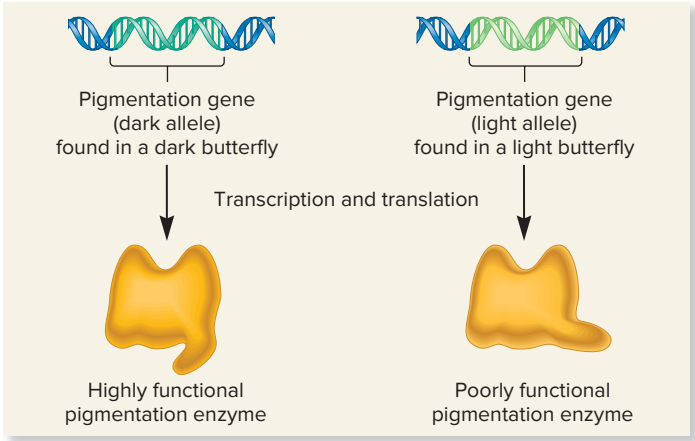
A complicated, yet very exciting, aspect of genetics is that our observations and theories span four levels of biological organization: molecules, cells, organisms, and populations. This broad scope can make it difficult to appreciate the relationship between

genes and traits. To understand this connection, we need to relate the following four phenomena:

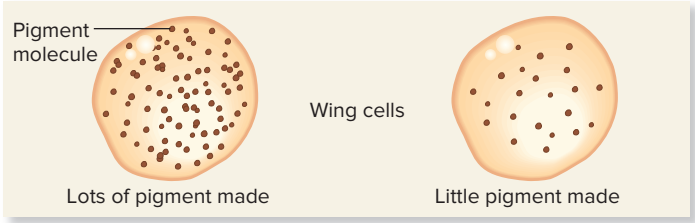
1. As we learned in Section 1.1, genes are expressed at the **molecular level.** In other words, gene transcription and translation lead to the production of a particular protein, which is a molecular process.
2. Proteins often function at the **cellular level.** The function of a protein within a cell affects the structure and workings of that cell.
3. An organism's traits are determined by the characteristics of its cells. We do not have microscopic vision, yet when we view morphological traits, we are really observing the properties of an individual's cells. For example, a red flower has its color because its cells make a red pigment. The trait of red flower color is an observation at the **organism level,** yet the trait is rooted in the molecular characteristics of the organism's cells.
4. A **species** is a group of organisms that maintains a distinctive set of attributes in nature. The occurrence of a trait within a species is an observation at the **population level.** Along with learning how a trait occurs, we also want to understand why a trait becomes prevalent in a particular species. In many cases, researchers discover that a trait predominates within a population because it promotes the reproductive success of the members of the population.

As a schematic example to illustrate the four levels of genetics, **Figure 1.7** shows the trait of pigmentation in a species of butterflies. One member of this species is light-colored and the other is very dark. Let's consider how we can explain this trait at the molecular, cellular, organism, and population levels.

1. At the molecular level, we need to understand the nature of the gene or genes that govern this trait. As shown in Figure 1.7a, a gene, which we will call the pigmentation gene, is responsible for the amount of pigment produced. The pigmentation gene can exist in two different forms called **alleles.** In this example, one allele confers a dark pigmentation and one causes a light pigmentation. Each of these alleles encodes a protein that functions as a pigment-synthesizing enzyme. However, the DNA sequences of the two alleles differ slightly from each other. This difference in the DNA sequence leads to a variation in the structure and function of the respective pigmentation enzymes.
2. At the cellular level (Figure 1.7b), the functional differences between the pigmentation enzymes affect the amount of pigment produced. The allele causing dark pigmentation, which is shown on the left, encodes an enzyme that functions very well. Therefore, when this gene is expressed in the cells of the wings, a large amount of pigment is made. By comparison, the allele causing light pigmentation encodes an enzyme that functions poorly. Therefore, when this allele is the only pigmentation gene expressed, little pigment is made.

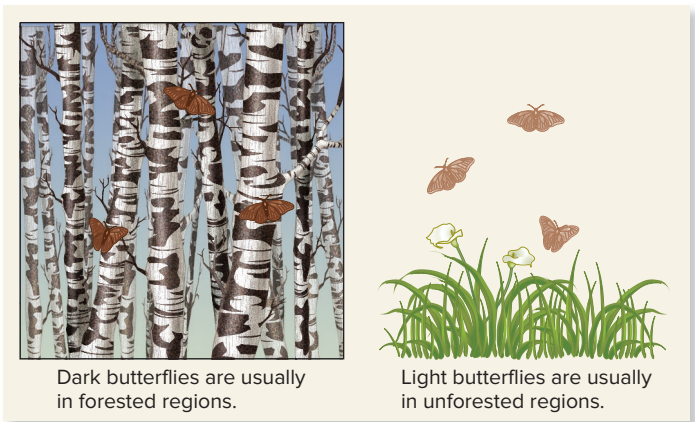


(a) Molecular level



(b) Cellular level

(c) Organism level



(d) Population level

FIGURE 1.7 **The relationship between genes and traits at the (a) molecular, (b) cellular, (c) organism, and (d) population levels.**

Concept Check: Which butterfly has a more active pigment-synthesizing enzyme, the light- or dark-colored one?

3. At the organism level (Figure 1.7c), the amount of pigment in the wing cells governs the color of the wings. If the pigment-synthesizing enzymes produce high amounts of pigment, the wings are dark-colored; if the enzymes produce little pigment, the wings are light.
4. Finally, at the population level (Figure 1.7d), geneticists want to know why a species of butterfly has some members with dark wings and others with light wings. One possible explanation is differential predation. The butterflies with dark wings might avoid being eaten by birds if they happen to live within the dim light of a forest. The dark wings would help to camouflage the butterfly if it were perched on a dark surface such as a tree trunk. In contrast, the light-colored wings would be an advantage if the butterfly inhabited a brightly lit meadow. Under these conditions, a bird might be less likely to notice a light-colored butterfly that was perched on a sunlit surface. A geneticist might study this species of butterfly and find that the dark-colored members usually live in forested areas and the light-colored members reside in unforested regions.

FIGURE 1.8 **Two dyeing poison frogs (*Dendrobates tinctorius*) showing different morphs within a single species.**

(a) ©Natalia Kuzmina/Shutterstock; (b) ©Valt Ahyppo/Shutterstock

Concept Check: Why do these two frogs look so different?

Inherited Differences in Traits Are Due to Genetic Variation

In Figure 1.7, we considered how gene expression can lead to variation in a trait of an organism, specifically, dark- versus light-colored wings in butterflies. Variation in traits among members of the same species is very common. For example, some people have black hair, and others have brown hair; some petunias have white flowers, but others have purple flowers. These are examples of **genetic variation.** This term describes the differences in inherited traits among individuals within a population.

In large populations that occupy a wide geographic range, genetic variation can be quite striking. Morphological differences have often led geneticists to misidentify two members of the same species as belonging to separate species. As an example, **Figure 1.8** shows two dyeing poison frogs that are members of the same species, *Dendrobates tinctorius.* They display dramatic differences in their markings. Such contrasting forms within a single species are termed **morphs.** You can easily imagine how someone might mistakenly conclude that these frogs are not members of the same species.

Changes in the nucleotide sequence of DNA underlie the genetic variation that we see among individuals. Throughout this textbook, we will routinely examine how variation in the genetic material results in changes in the outcome of traits. At the molecular level, genetic variation can be attributed to different types of modifications.

- Small or large differences can occur within gene sequences. When such changes initially occur, they are called **gene mutations,** which are heritable changes in the genetic material. Gene mutations result in genetic variation in which a gene is found in two or more alleles, as previously described in Figure 1.7. In many cases, gene mutations alter the expression or function of a protein that a gene specifies.
- Major alterations can also occur in the structure of a chromosome. A large segment of a chromosome can be lost, rearranged, or reattached to another chromosome.
- Variation may also occur in the total number of chromosomes. In some cases, an organism may inherit one too many or one too few chromosomes. In other cases, it may inherit an extra set of chromosomes.

Variations within the sequences of genes are a common source of genetic variation among members of the same species. In humans, familiar examples of sequence variation involve genes for eye color, hair texture, and skin pigmentation. Chromosome variation—a change in chromosome structure or number (or both)—is also found, but this type of change is often detrimental. Many human genetic disorders are the result of chromosomal alterations. An example is Down syndrome, which is due to the presence of an extra chromosome (**Figure 1.9a**). By comparison, chromosome variation in plants is common and often results in plants with superior characteristics, such as increased resistance to disease. Plant breeders have frequently exploited this observation. Cultivated varieties of wheat, for example, have many more chromosomes than the wild species (**Figure 1.9b**).

Traits Are Governed by Genes and by the Environment

In our discussion thus far, we have considered the role that genes play in the outcome of traits. Another critical factor is the **environment**—the surroundings in which an organism exists. A variety of factors in an organism's environment profoundly affect its morphological and physiological features. For example, a person's diet greatly influences many traits, such as height, weight, and even intelligence. Likewise, the amount of sunlight a plant receives affects its growth rate and the color of its flowers. The term **norm of reaction** refers to the effects of environmental variation on an individual's traits.

(a) (b)

FIGURE 1.9 Examples of chromosome variation. (a) A person with Down syndrome. She has 47 chromosomes rather than the common number of 46, because she has an extra copy of chromosome 21. **(b)** A wheat plant. Bread wheat is derived from the contributions of three related species with two sets of chromosomes each, producing an organism with six sets of chromosomes.

Concept Check: Are these examples of gene mutations, variation in chromosome structure, or variation in chromosome number?

FIGURE 1.10 Environmental influence on the outcome of PKU. This girl with PKU has developed normally because she followed a diet that is very low in phenylalanine.

Concept Check: What would have been the consequences if this girl had followed a standard diet, which contains a higher amount of phenylalanine?

External influences may dictate the way that genetic variation is manifested in an individual. An interesting example is the human genetic disease **phenylketonuria (PKU).** Humans have a gene that encodes an enzyme known as phenylalanine hydroxylase. Most people have two functional copies of this gene. People with one or two functional copies of the gene can eat foods containing the amino acid phenylalanine and metabolize it properly.

A rare variation in the sequence of the phenylalanine hydroxylase gene results in a nonfunctional version of this protein. Individuals with two copies of this rare, inactive allele cannot metabolize phenylalanine properly. Such individuals represent about 1 in 8000 births in the United States. When given a standard diet containing phenylalanine, individuals with this disorder are unable to break down this amino acid. Phenylalanine accumulates and is converted into phenylketones, which are detected in the urine. PKU individuals manifest a variety of detrimental traits, including mental impairment, underdeveloped teeth, and foul-smelling urine. In contrast, when PKU individuals are identified at birth and raised on a restricted diet that is low in phenylalanine, they develop normally (**Figure 1.10**). Fortunately, through routine newborn screening, most affected babies in the United States are now diagnosed and treated early. PKU provides a dramatic example of how the environment and an individual's genes can interact to influence the traits of the organism.

During Reproduction, Genes Are Passed from Parent to Offspring

Now that we have considered how genes and the environment govern the outcome of traits, we can turn to the issue of inheritance. How are traits passed from parents to offspring? The foundation for our understanding of inheritance came from the studies of pea plants by Gregor Mendel in the nineteenth century. His work revealed that genetic determinants, which we now call genes, are passed from parent to offspring as discrete units. We can predict the outcome of many genetic crosses based on Mendel's laws of inheritance.

The inheritance patterns identified by Mendel can be explained by the existence of chromosomes and their behavior during cell division.

- Like Mendel's pea plants, sexually reproducing species are commonly **diploid.** This means that their cells contain two copies of each chromosome, one from each parent. The two copies are called **homologs** of each other.
- Because genes are located within chromosomes, diploid organisms have two copies of most genes. Humans, for example, have 46 chromosomes, which are found in homologous pairs (**Figure 1.11a**). With the exception of the sex chromosomes (X and Y), each homologous pair contains the same kinds of genes. For example, both copies of human chromosome 12 carry the gene that encodes phenylalanine hydroxylase, which was discussed previously. Therefore, an individual has two copies of this gene that may or may not be identical alleles.
- Most cells of the human body that are not directly involved in sexual reproduction contain 46 chromosomes. These cells are called **somatic cells.** In contrast, the **gametes**—sperm and egg cells—contain half that number (23) and are termed **haploid** (**Figure 1.11b**).
- The union of gametes during fertilization restores the diploid number of chromosomes. The primary advantage of sexual reproduction is that it enhances genetic variation. For example, a tall person with blue eyes and a short person with